

### Claims

1. A nucleic acid molecule comprising a 5' portion of an intestinal lactase-phlorizine hydrolase (LPH) gene contributing to or indicative of adult-type hypolactasia wherein said nucleic acid molecule is selected from the group consisting of
  - (a) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO: 1, the sequence of SEQ ID NO:1 is also depicted in Fig. 4 and comprised in the sequence as depicted in Fig. 8;
  - (b) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO: 2, the sequence of SEQ ID NO:2 is also depicted in Fig. 5 and comprised in the sequence as depicted in Fig. 9;
  - (c) a nucleic acid molecule of at least 20 nucleotides the complementary strand of which hybridizes under stringent conditions to the nucleic acid molecule of (a) or (b), wherein said polynucleotide/nucleic acid molecule has at a position corresponding to position -13910 5' from the LPH gene a cytosine residue; and
  - (d) a nucleic acid molecule of at least 20 nucleotides the complementary strand of which hybridizes under stringent conditions to the nucleic acid molecule of (a) or (b), wherein said polynucleotide/nucleic acid molecule has at a position corresponding to position -22018 5' from the LPH gene a guanine residue.
2. A nucleic acid molecule comprising a 5' portion of an intestinal lactase-phlorizine hydrolase (LPH) gene wherein said nucleic acid molecule is selected from the group consisting of
  - (a) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO:3, the sequence of SEQ ID NO:3 is also depicted in Fig. 6;
  - (b) a nucleic acid molecule having or comprising the nucleic acid sequence of SEQ ID NO:4, the sequence of SEQ ID NO:4 is also depicted in Fig. 7;
  - (c) a nucleic acid molecule the complementary strand of which hybridizes

under stringent conditions to the nucleic acid molecule of (a) or (b), wherein said polynucleotide/nucleic acid molecule has at a position corresponding to position -13910 of the LPH gene a thymidine residue; and

- 5 (d) a nucleic acid molecule the complementary strand of which hybridizes under stringent conditions to the nucleic acid molecule of (a) or (b), wherein said polynucleotide/nucleic acid molecule has at a position corresponding to position -22018 of the LPH gene a adenosine residue.
- 10 3. The nucleic acid molecule of claim 1 or 2 which is genomic DNA.
4. The nucleic acid molecule of claim 3 wherein said genomic DNA is part of a gene.
- 15 5. A fragment of the nucleic acid molecule of any one of claims 1 to 4 having at least 14 nucleotides wherein said fragment comprises nucleotide position -13910 or nucleotide position -22018 of the LPH gene.
- 20 6. A nucleic acid molecule which is complementary to the nucleic acid molecule of any one of claims 1 and 3 to 5.
7. A nucleic acid molecule which is complementary to the nucleic acid molecule of any one of claims 2 to 5.
- 25 8. A vector comprising the nucleic acid molecule of any one of claim 1 and 3 to 5.
9. A vector comprising the nucleic acid molecule of any one of claims 2 to 4.
- 30 10. A primer or primer pair, wherein the primer or primer pair hybridizes under stringent conditions to the nucleic acid molecule of any one of claims 1 and 3 to 5 comprising nucleotide position -13910 or -22018 of the LPH gene or to the complementary strand thereof.

11. A primer or primer pair, wherein the primer or primer pair hybridizes under stringent conditions to the nucleic acid molecule of any one of claims 2 to 5 comprising nucleotide position -13910 or -22018 of the LPH gene or to the complementary strand thereof.
12. A non-human host transformed with the vector of claim 6.
13. A non-human host transformed with the vector of claim 7.
14. The non-human host of claim 12 or 13 which is a bacterium, a yeast cell, an insect cell, a fungal cell, a mammalian cell, a plant cell, a transgenic animal or a transgenic plant.
15. An antibody or aptamer or phage that specifically binds to the wild-type nucleic acid molecule of any one of claims 1 and 3 to 6 but not to the corresponding wild-type nucleic acid molecule
16. An antibody or aptamer or phage that specifically binds to the wild-type nucleic acid molecule of any one of claims 2 to 5 and 7 but not to the corresponding mutant sequence contributing to or indicative of adult-type hypolactasia.
17. A pharmaceutical composition comprising the wild-type nucleic acid molecule of claim 2, 3, 4 or the vector of claim 9.
18. A diagnostic composition comprising the nucleic acid molecule of any one of claims 1 to 7, the vector of claim 8 or 9, the primer or primer pair of claim 11 or 12, and/or the antibody aptamer and/or phage of claim 15 or 16.
19. A method for testing for the presence or predisposition of adult-type hypolactasia or associated trait comprising testing a sample obtained from a prospective patient or from a person suspected of carrying such a predisposition for the presence of the nucleic acid molecule of any one of claims 1 and 3 to 6 in a homozygous or heterozygous state.

20. A method for testing for the presence or predisposition of adult-type hypolactasia or associated trait comprising testing a sample obtained from a prospective patient or from a person suspected of carrying such a predisposition for the presence of the nucleic acid molecule of any one of claims 2 to 5 and 7 in a homozygous or heterozygous state.
21. The method of claim 19 or 20, wherein said testing comprises hybridizing the complementary nucleic acid molecule of claim 6 which is complementary to the nucleic acid molecule contributing to or indicative of adult-type hypolactasia or the nucleic acid molecule of claim 7 which is complementary to the wild-type sequence as a probe under stringent conditions to nucleic acid molecules comprised in said sample and detecting said hybridization.
22. The method of any one of claims 19 or 21 further comprising digesting the product of said hybridization with a restriction endonuclease or subjecting the product of said hybridization to digestion with a restriction endonuclease and analyzing the product of said digestion.
23. The method of claim 21, wherein said probe is detectably labeled.
24. The method of claim 19 or 20, wherein said testing comprises determining the nucleic acid sequence of at least a portion of the nucleic acid molecule of any one of claims 1 to 7, said portion comprising nucleotide position -13910 and/or nucleotide position -22018 of the LPH gene.
25. The method of claim 24, wherein the determination of the nucleic acid sequence is effected by solid-phase minisequencing.
26. The method of claim 24 further comprising, prior to determining said nucleic acid sequence, amplification of at least said portion of said nucleic acid molecule.

27. The method of claim 19 or 20, wherein said testing comprises carrying out an amplification reaction wherein at least one of the primers employed in said amplification reaction is the primer of claim 10 or belongs to the primer pair of claim 10, comprising assaying for an amplification product.
- 5 28. The method of claim 19 or 20, wherein said testing comprises carrying out an amplification reaction wherein at least one of the primers employed in said amplification reaction is the primer of claim 11 or belongs to the primer pair of claim 11, comprising assaying for an amplification product.
- 10 29. The method of any one of claims 26 to 28 wherein said amplification is effected by or said amplification is the polymerase chain reaction (PCR).
- 15 30. A method for testing for the presence or predisposition of adult-type hypolactasia comprising assaying a sample obtained from a human for specific binding to the antibody or aptamer or phage of claim 15
- 20 31. A method for testing for the presence or predisposition of adult-type hypolactasia comprising assaying a sample obtained from a human for specific binding to the antibody or aptamer or phage of claim 16.
32. The method of claim 30 or 31, wherein said antibody or aptamer or phage is detectably labeled.
- 25 33. The method of any one of claims 30 to 32, wherein the test is an immunoassay.
- 30 34. The method of any one of claims 19 to 33, wherein said sample is blood, serum, plasma, fetal tissue, saliva, urine, mucosal tissue, mucus, vaginal tissue, fetal tissue obtained from the vagina, skin, hair, hair follicle or another human tissue.

35. The method of any one of claims 19 to 34, wherein said nucleic acid molecule from said sample is fixed to a solid support.
- 5 36. The method of claim 35, wherein said solid support is a chip, a silica wafer, a bead or a microtiter plate.
37. Use of the nucleic acid molecule of any one of claims 1 to 7 for the analysis of the presence or predisposition of adult-type hypolactasia.
- 10 38. Kit comprising the nucleic acid molecule of any one of claims 1 to 7, the primer or primer pair of claim 11 or 12, the vector of claim 8 or 9, and/or the antibody aptamer and/or phage of claim 15 or 16 in one or more containers.
- 15 39. Use of the nucleic acid molecule of any one of claims 2 to 4 or the vector of claim 7 in gene therapy.
40. The use of claim 39, wherein said gene therapy treats or prevents adult-type hypolactasia.